NATIONAL CENTRE FOR INHERITED METABOLIC DISORDERS (NCIMD)

EMERGENCY LETTER FOR HOSPITAL – Urea Cycle Disorders

(For use in patients with Ornithine Transcarbamylase (OTC) Deficiency, Arginosuccinic Aciduria (ASA), Citrullinaemia Type 1, or Carbamyl Phosphate Synthetase (CPS1) Deficiency)

Addressograph

AT RISK OF ACUTE METABOLIC CRISIS
Please attend to this patient without delay
Always contact the NCIMD for further advice

DIAGNOSIS

Urea cycle disorders (UCDs) are inherited disorders of protein metabolism. The body is unable to correctly remove nitrogen via the urea cycle, resulting in a build-up of ammonia. Ammonia is a neurotoxic compound that causes cerebral oedema, encephalopathy, coma and death.

Treatment of urea cycle disorders is with a low protein diet, and with medications known as nitrogen scavengers (sodium benzoate, sodium phenylbutyrate or glycerol phenylbutyrate) to reduce ammonia production. Patients may be on a measured amount of synthetic protein. They may also receive supplements of the amino acids arginine or citrulline, depending on the type of UCD.

Signs of decompensation in UCDs include **irritability, lethargy, vomiting, poor feeding, confusion, slurred speech, seizures and ataxia.** When a patient with a UCD is under metabolic stress e.g. they are unwell, fasting for a prolonged period of time, have excessive protein intake, have surgery, or sustain a fracture or other trauma, they are at high risk of decompensation. It is critical to react early to any signs of illness by implementing their unwell plan.

NOTES FOR PARENTS/CARERS

Contact the metabolic team if your child becomes unwell to discuss your child's care. Metabolic Clinical Nurse Specialist contact: (01) 878 4409 (during working hours).

If you are advised to attend the emergency department, remember to bring your medications and feeding supplies, and a copy of this letter and your emergency diet plan with you.

EMERGENCY MANAGEMENT

Always take illness in a patient with a urea cycle disorder seriously. Always discuss this patient with the metabolic consultant on call without delay.

- **Stop natural protein** (unless advised otherwise by metabolic team)
- Discuss with the metabolic team if the synthetic protein recipe should be given
- Assess and resuscitate as necessary as per APLS protocols
- Obtain IV access and send bloods:
    - glucose and ketones (point of care)
    - FBC, renal/liver/bone profile, venous blood gas, ammonia, glucose, plasma amino acids
    - Other tests as indicated by presentation (blood cultures, urine culture, NPA etc)
- Start 10% dextrose at 120% maintenance rate with 0.45% saline and 2 mmol/kg/day of potassium chloride (KCl can be added once you have confirmed that urine has been passed), adjusted depending on electrolyte levels
- Start IV 20% Lipid (Intralipid or SMOF) 2-3 g/kg/day to prevent catabolism (in discussion with the metabolic team)
- Provide additional calories as tolerated (see emergency diet plan) via enteral route (PO/NG/PEG)
- If child is nauseated or vomiting, give ondansetron
- Manage fever proactively with paracetamol or ibuprofen
- Keep **detailed and accurate** record of all diet and fluid intake
- If infection suspected, manage in line with the hospital sepsis pathway as patients can decompensate quickly. Consider discussion with Micro/ID especially in cases of sepsis or if the patient has multi-drug resistant organisms
- Depending on the initial ammonia level, the metabolic consultant may recommend either an emergency IV regimen with sodium benzoate, sodium phenylbutyrate and arginine, OR an increase in the usual oral medication regimen.
- Due to the amount of sodium present in sodium benzoate and sodium phenylbutyrate, electrolytes should be monitored closely. If using IV preparations, the fluids used for calorie provision (described above at 120% maintenance) may need to be adjusted depending on electrolyte levels to reduce the sodium content
- Patients with ASA in particular can develop profound hypokalaemia and may need considerable amounts of IV potassium replacement, adjusted depending on blood levels
- **This plan is for the immediate management only and continued management must be discussed with the metabolic team**

The on call service for the National Centre for Inherited Metabolic Disorders is available to discuss this patient at +353 1 878 4200 via the CHI at Temple Street switchboard (available 24 hours a day). The metabolic ward nurse is also available to discuss at +353 1 878 4200, ask for bleep 836 (available 24 hours a day).